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Brief Clinical Report

Elejalde Syndrome: A Case Report

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Elejalde syndrome is a rare condition comprising a high birth weight, swollen globular body, a short neck with redundant skin folds, postaxial polydactyly, omphalocele, enlarged liver and kidneys, and renal dysplasia. The syndrome, also known as acrocephalopolydactylous dysplasia, is thought to be an autosomal recessive trait. We report on a liveborn infant with this condition. Am. J. Med. Genet. 69:406-408, 1997. © 1997 Wiley-Liss, Inc.

KEY WORDS: Elejalde syndrome; acrocephalopolydactylous dysplasia; autosomal recessive

INTRODUCTION

Elejalde et al. [1977] described two sibs born to consanguineous parents. Both of them showed a massive increase in birth weight, a swollen globular body with thick skin, apparently short limbs, polydactyly, craniosynostosis with acrocephaly, omphalocele, and abnormal face. Autopsy documented abdominal organomegaly, ascites, and cystic renal dysplasia, with excessive amounts of connective tissue and perivascular proliferation of nerve fibers in many organs.

Lurie et al. [1991], in a wide discussion of cerebroreno-digital syndromes, describes a further case of Elejalde syndrome although in little detail. Nevin et al. reported an 18-week fetus with Elejalde syndrome in 1994. Here we present a neonate born at 24 weeks of gestation who closely resembles case 2 in the original paper of Elejalde et al. Although the present case and that of Nevin et al. were born in Northern Ireland, the families are apparently unrelated.

CLINICAL REPORT

This infant was born to a 32-year-old para 2 woman with two normal children. There is no history of ma-

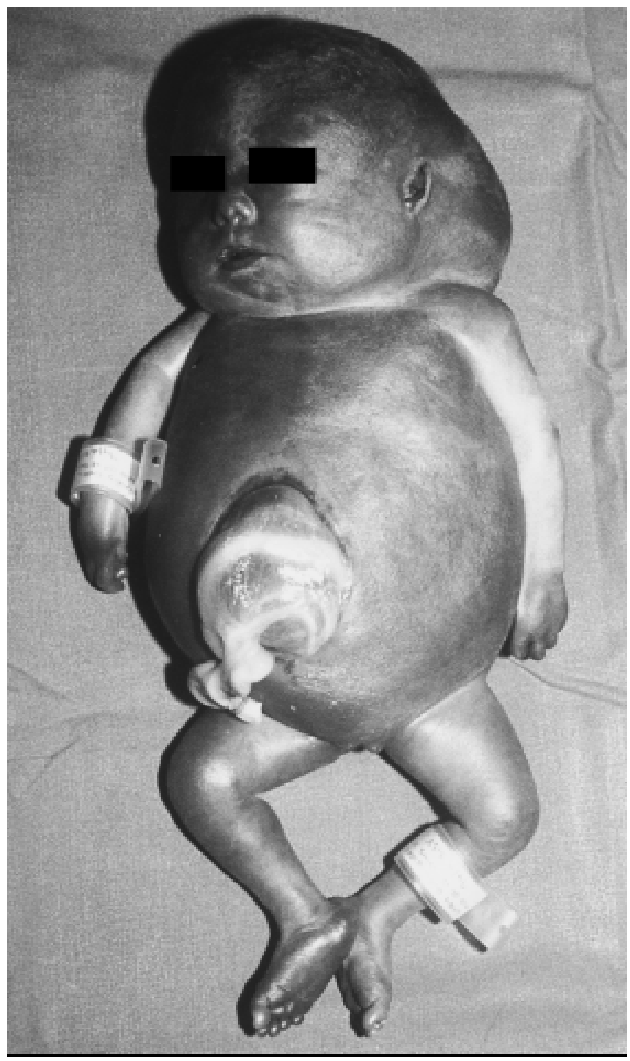

Fig. 1. Baby with Elejalde syndrome: swollen globular body, short neck, omphalocele, and apparent shortness of limbs.

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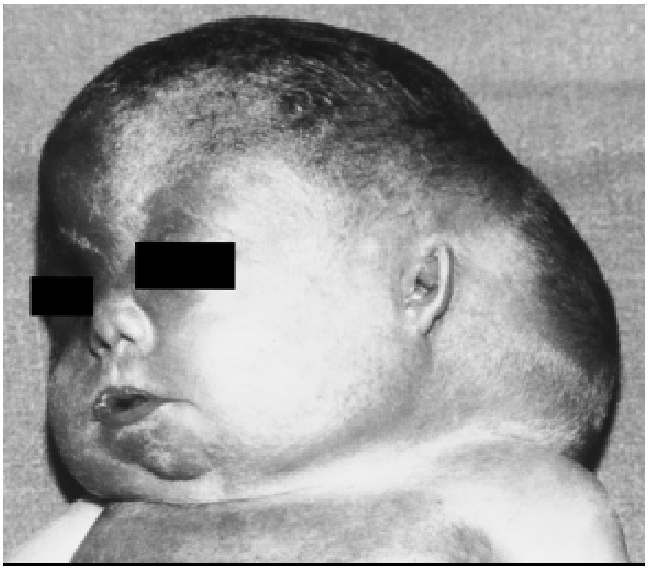

Fig. 2. Facial appearance in Elejalde syndrome: hypertelorism, epicanthic folds, small flat nose, and abnormally modeled ears.

ternal illness and the parents are nonconsanguineous. The mother developed marked polyhydramnios and an ultrasound scan showed a large-for-date fetus with probable hydrops fetalis, cystic hygroma, and omphalocele. Amniocentesis was performed and the fetal karyotype was found to be 46,XX. The child was born at 24 weeks gestation but died at age 2 hours.

A limited autopsy was performed, the internal examination being confined to the abdominal cavity. The infant weighed 1,500 g and the placenta 390 g, both almost twice the expected weight for the gestational age. The body measurements were as follows: head circumference 30.6 cm, crown-rump length 24 cm, crown heel length 35 cm, and foot length 4.9 cm. The body had a globular appearance with a large swollen head, short thick neck, broad chest, and protruberant abdomen (Fig. 1). The limbs were short and there was an omphalocele containing loops of bowel. The skin was thick, shiny, and taut. This thickness was most prominent over the face and head where it had resulted in the formation of a soft tissue mass over the posterior scalp and the nape of the neck.

Examination showed small abnormally modeled ears, hypertelorism, epicanthic folds, and slightly slanted palpebral fissures. The nose was small and flat and the mouth and lower jaw appeared small (Fig. 2).

The abdominal protruberance was due in part to ascites and in part to organomegaly, the liver, spleen, and kidneys being enlarged. The intestines appeared normal apart from the loops of small bowel within the omphalocele. A small accessory spleen was noted at the hilum of the spleen. There was no evidence of cyst formation within the kidneys and the bladder appeared normally formed.

Histological abnormalities were noted in several organs. The kidneys did not show capsular fibrosis or the features of cystic dysplasia but there did appear to be glomerular crowding with a reduction in the numbers of intervening tubules. Some of the glomeruli also appeared enlarged and a small number was sclerotic. Within the liver there was expansion of portal tracts by fibrous tissue which extended to connect adjacent portal tracts giving the liver a rather nodular appearance. There was a ductal plate malformation. Well-formed bile ductules were absent from most portal tracts whilst the ductal plates were irregular and spiky. The pancreas showed a mild degree of interstitial fibrosis and nerve trunks appeared somewhat prominent within the fibrous tissue (Fig. 3). Skin from the abdominal wall showed markedly thickened subcutis composed of abundant loose collagenous tissue contain-

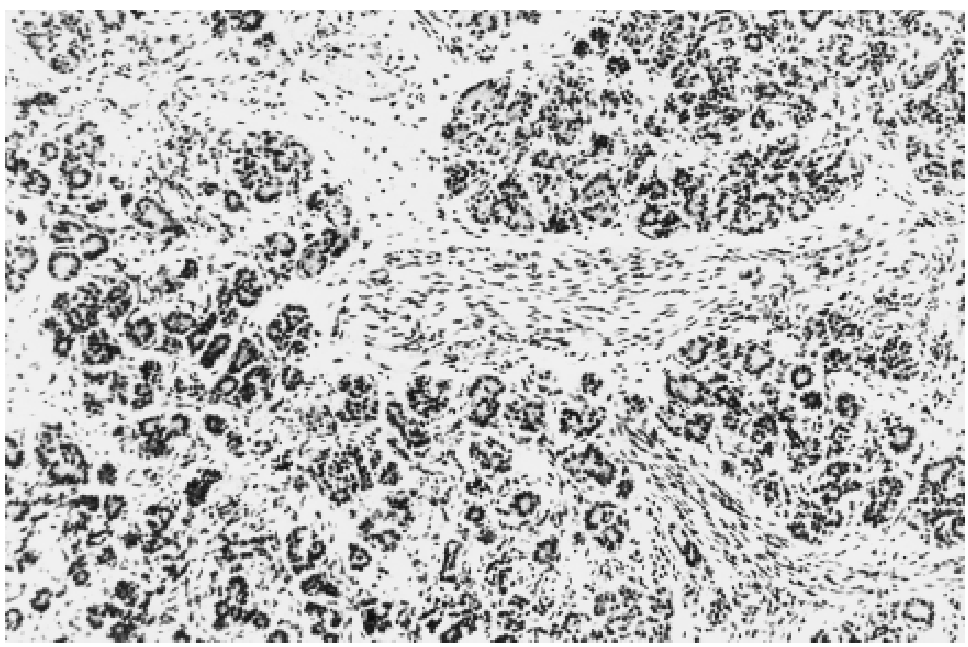

Fig. 3. Pancreas showing prominent nerve trunks.

TABLE I. Macroscopic Abnormalities in Recorded Cases of Elejalde Syndrome*

Macroscopic findings	Elejalde et al. 1	Elejalde et al. 2	Lurie et al.	Nevin et al.	Present case
Increased birth weight	+	+	+	+	+
Increased placental weight	+	+	NR	+	+
Swollen globular body	+	+	+	+	+
Apparent micromelia	+	+	NR	+	+
Short neck, skin folds	++	++	++	++	++
Thick skin	+	+	NR	NR	+
Craniosynostosis/acrocephaly	+	+	NR	+	−
Slanted palpebral fissures	+	+	NR	NR	+
Hypertelorism, epicanthic folds	++	++	NR	NR	++
Dysplastic ears	+	+	NR	+	+
Small nose	+	+	NR	+	+
Polydactyly	−	+	+	+	−
Omphalocele, ascites	++	+NR	−NR	−NR	++
Enlarged liver	+	+	NR	NR	+
Enlarged kidneys, cystic kidneys	++	+,+	−,+	NR,NR	+−
Splenic abnormality	−	+	+	+	+
Hypoplastic lungs	+	+	+	NR	NE
Cytogenetics	NE	46XX	NR	46XY	46XX

*NR, not recorded; NE, not examined.

ing numerous small blood vessels. Cuffs of loose mesenchyme surrounded a number of the blood vessels and with these were associated slightly prominent nerve fibers. The underlying skeletal muscle appeared unremarkable, as did the spleen, duodenum, and ovary. Postmortem radiologic study was performed but showed no evidence of craniosynostosis, acrocephaly, shortness of limbs, or micrognathia.

DISCUSSION

The manifestations and the findings in the four previously reported cases and the present case of Elejalde syndrome [McKusick 200995] are summarized in Table I. The external appearance of our case closely resembles that of case 2 of Elejalde et al. although there was no evidence of craniosynostosis or polydactyly. Postmortem radiologic study showed the shortness of the limbs and the micrognathia to be apparent rather than real, presumably as a result of the swollen head and abdomen.

It would appear that there are similarities in the internal and histological findings but also some differences. We found no evidence of cystic change in the kidneys although they showed deranged development. The liver showed portal tract fibrosis and a ductal plate malformation, but no proliferation of bile canaliculi. The interstitial fibrosis and perivascular nerve trunk proliferation described by Elejalde et al. were present, but only to a mild degree in the pancreas.

Elejalde et al. demonstrated an increased rate of fibroblast proliferation in cell culture, and it has been suggested that the condition is in fact an overgrowth syndrome.

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